April 1899

THESIS.

on

THE PROGRESSIVE MUSCULAR DYSTROPHIES.

Illustrated by two cases.

presented by

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THE PROGRESSIVE MUSCULAR DYSTROPHIES.

Of late years the attention of Neurologists has been directed to a class of nervous diseases which has, for its predominant symptom, a progressive wasting of the muscles of the body. Aran and Duchenne first described Progressive Muscular Atrophy, and they published a series of cases in which the atrophy, commencing in the small muscles of the hands, gradually spread through the muscles of the upper extremities. The lesion, in these cases, is undoubtedly situated in the Anterior Cells of the Spinal Grey matter; that is, the cases are true myelopathies, and the disease is now known as Anterior Poliomyelitis Chronica. But since that day many cases have been described which unfortunately do not possess any such definite Pathology, and which, although their chief characteristic is Muscular wasting, do not, either clinically or pathologically, resemble in any marked degree the above mentioned disease. This group of cases, then, which for want of a better title have been collected together under the name of Progressive Muscular Dystrophy, although presenting many features in common, yet vary from one another in such a way that our present

knowledge can not give them a common Pathological cause.

Some of the cases seem to be explained by a lesion in the Anterior horns of the Spinal Grey Matter, that is, are Myelopathic in origin; others seem rather to be caused by changes in the peripheral nerves supplying the affected muscles, that is, are Neuropathic in origin; while yet again there are further cases in which no lesion of any part of the nervous system can be made out, and so far we have to look for an explanation of the Muscular Wasting, to the changes in the affected muscles themselves, that is, these cases are myopathic in origin. It will be my endeavour, in this Thesis, to review, as far as possible, the work published in connection with this class of nervous diseases, and to attempt to form some classification of them from the Clinical and Pathological evidence we possess, as well as to bring before your notice two cases not hitherto described. These two cases, although not shedding any fresh light on the pathological side of the disease, are of great interest on account of their clinical symptoms, more especially as they resemble very closely in their character cases already published, and it is only by

careful study of these cases, together, that we can hope to make a forecast as to what is the primary cause of the disease, a cause which in due season, as our knowledge advances and our methods of investigation improve, no doubt will stand plain and distinct before us.

The writings of Erb, Charcot, Duchenne, Shultze, Landouzy, and others first drew attention to these peculiar forms of muscular wasting. Erb pointed out that the various types described by these observers could be grouped together into what he called one "Nosological Species," and he proposed for this the name which is still in use "Progressive Muscular Dystrophy." He was extremely cautious as to the cases he admitted under this head, and, in fact, refused to consider that the "Peroneal type," described by Tooth and Marie,came within its scope. More recent writers have wisely admitted its claim for a place (at present) among the Progressive Muscular Dystrophies. Erb,[1] in a most interesting lecture on the Muscular Dystrophies, stated that, as far as he could judge, the changes in the muscles in all the various forms of dystrophy were, in all essential points, alike, and then proceeded to give a sketch of what he thought

1. [illegible]

might be the true cause of these changes, and where the lesion occasioning them might be looked for. He does this in such an extremely interesting and prophetic way that I may be excused if I quote his words in full. "This is the place to say a little about the proper nature, pathogenesis, and exact seat of the lesion. It is not necessary to prove further that it is neither a simple atrophy of the muscles nor an inflammatory affection with its consequences; there is unquestionably a more complicated disturbance of nutrition, regarding the nature of which, for the present, I would rather not express a definite opinion. Pathological anatomy has led many to consider it simply as a local muscular affection, quite independent of the central nervous system, a pure myopathy. But a good deal of doubt has been expressed (for example by Knoll) regarding this view, and I myself, in my earlier work, brought forward some reasons which prevented me from accepting it without more inquiry. A much larger experience, a good deal of consideration given to the subject, and most of all the results of the previously mentioned autopsies have confirmed me in my view. The considerations which weigh with me are various. The muscles depend for their nutrition to a very large

extent on trophic nerve centres; the localisation of this atrophy frequently follows in a noticeable way the exact course of the nerves in a plexus, or the disposition of the centres in the central organ, and occasionally we find a case of spinal amyotrophy (Strumpell's case) presenting an almost exactly similar arrangement. Hereditary influence is of great importance; mental aberrations are common among the patients and other neuroses frequently occur in their families. Further, even in the undoubtedly spinal cases, such as acute anterior poliomyelitis similar morbid changes (hypertrophy, proliferation of nuclei, division of the fibres) both in the muscles and connective tissue have been pointed out by W. Müller, Déjérine-Huet , Joffroy-achard, and Hitzig. In pseudo-hypertrophic cases, malformations and changes of a minor kind in the spinal cord have been met with. When I consider these facts, and bear in mind further the results of Henbuer's and Frohmaier's observations I cannot avoid the suspicion that after all the affection may be dependent on the nervous system. It is tempting to suppose, as I formerly expressed it, that we have to do with a kind of trophoneurosis, having its origin in the trophic centres of the cord - a disturbance

of the function of these centres which finds its expression in the very complicated muscle changes of the disease. While on this supposition there are, as a rule, no coarse nerve changes, now and then, and after the affection has lasted a long time or been very intense, such a change does become visible. The idea is inevitable that if something like this is the case the relations between dystrophy and spinal amyotrophy will turn out again in the end to be close and intimate. The latter would represent an affection of the trophic centres that from the very first is a distinct coarse anatomical lesion taking effect in a degenerative atrophy of the muscles with fibrillar twitchings, reaction of degeneration, &c; the former at the outset would be merely a functional disturbance of these centres, conditioned probably by different causes, and expressing itself as muscular dystrophy with all its characteristic symptoms. At the same time there would remain the possibility that even this merely functional disturbance might in the long run become associated with a coarse lesion of the centres. Many things about these affections would agree very well with such a supposition, among them the occasional occurrence of the reaction of degeneration in dystrophic

cases and the occasional initial localisation of spinal amyotrophy in the shoulder and trunk. But I will not spin out this discussion to any greater length; the whole question is by no means yet ready for decision, and the future alone can lift the veil and reveal the finer processes that as yet lie hid from us."

Erb clearly feels that the explanation of the muscular changes in these cases of Progressive Muscular Dystrophy will be found to consist in some central nervous lesion, but his difficulty was to bring forward sufficient evidence to prove it. That difficulty, unfortunately, remains with us still, and, although several post mortem examinations have been made of late years on cases falling under the title of Progressive Muscular Dystrophy, our present methods of Pathological research failed, in some of them, to reveal any changes in the nervous system. On the other hand the results obtained by the microscopical examination of the nerves and spinal cord by some observers give great encouragement to holders of the theory that the cause of the disease is after all a tropho-neurosis.

I propose to give here a short account of the chief clinical features of the various types of Progressive Muscular Dystrophy, and will then proceed to enumerate

the various types, and consider what light recent research has thrown on their obscure pathology. That being done, it remains to be seen if some kind of classification cannot be arrived at, based on our present clinical and pathological knowledge.

It is satisfactory to think that we can clinically, if not pathologically, recognise examples of the various types as being closely related to each other. The physical signs of the disease in each case being so much alike, as follows:- The onset is always insidious and gradual, weakness and wasting, as a rule, first call the victim's attention to his condition. The weakness is proportionate to the amount of wasting. This wasting is progressive, spreading from muscle to muscle, and from limb to limb. The wasting may start simultaneously and symmetrically on the two sides and progress so; or one side may be affected first and later the other side become involved. Along with this atrophy of muscle there may be some hypertrophy (true or false), though this cannot always be observed. There is usually a marked hereditary tendency in the disease, and frequently several members of a family are affected. Another striking feature is that the disease usually commences in childhood, or early adult

life. The functions of the nervous system, other than the motor, are unaltered; sensory symptoms are rarely present, and the sphincters are unaffected. Fibrillary twitchings in the affected muscles seldom are seen, though they sometimes occur in the "Peroneal type" of the disease, and did so in the case I am about to describe. Electrical irritability of the muscles is lessened in proportion to the wasting, and this is true both for the Faradic and Galvanic currents. The reaction of degeneration is not in every case obtained.

Recent writers more or less agree in considering the following to be the chief types of the muscular dystrophies.

I. Pseudo-hypertrophic paralysis.
II. The "Juvenile form" of Erb.
III. The "Facio-scapulo-humeral" form.
IV. The Hereditary form of Leyden.
V. The Atrophic form of myopathic muscular atrophy.
VI. The Diffuse form of muscular atrophy of infancy and early childhood.
VII. The Peroneal type.

Pseudo-hypertrophic paralysis was first described by Duchenne[2], who held that the disease was a primary affection of the muscles. Billroth, later, showed that the hypertrophy was only apparent and not real. Erb[3] described the muscle changes as consisting of a considerable hypertrophy of the fibres, of all

possible degrees of atrohpy of the fibres, of increase of the nuclei both at the edges and in the interior, of vacuolation, sometimes to a considerable extent, of increase and overgrowth of the connective tissue, and lastly of lipomatosis of this overgrowth. Most careful study and examination of the nervous system at post mortems, by several observers, has revealed no pathological changes likely to be the cause of the affection. Dr. Gowers, holding that"the essential lesion is the growth of connective tissue by which the muscular fibres are damaged, whether fatty tissue is found or not." This was the state of our knowledge up till 1894, when Von Babes published (in La Roumaine Medicale, Aug. 1894) the result of his examination of the nerves from a case of Pseudo-hypertrophic paralysis, and one of Juvenile Muscular atrophy. He found the motor fibre going to the muscle thin and attenuated, and the terminal plate of small size (atrophied or non-developed) and much simplified. Another specimen showed not only an atrophied condition of the terminal plate, but the presence of small fusiform cells about this region. These cells have been shown to be present in the experimental degeneration (Wallerian) induced in the nerve terminations by section of the nerve trunk. He suggested that there was a defective

development of the nerve ending and of the fibre which gave rise to it, and, as a result, there was degeneration (dissolution) of the already formed end plate. These most interesting observations still await confirmation by other investigators, but, if they are to be found in all cases of Pseudo-hypertrophic paralysis, it is probable that, eventually, some central lesion will be made out which gives rise to these peripheral nerve changes, and so causes the complicated alterations found in the affected muscles.

The "Juvenile form" was first recognised by Erb(4) in 1884; he based his description of the type on clinical evidences, and considered the atrophy to be due to primary changes in the muscles themselves. Dreschfeld(5) confirms this view by a post mortem examination made on a case of Erb's Paralysis, in which he found the brain, spinal cord, the intra-muscular and large nerves, to be quite normal. Singer(6), on the other hand, gives an account of a case which seems to belong to this type. It was that of a woman aged 34, with no heredity of a neurosis, who suffered from weakness of the lower extremities, with some hypertrophy of the calf muscles. The muscles of the shoulder girdle were much atrophied, but the small muscles of

the hand were unaffected. He found, microscopically, the usual appearances in the affected muscles, but also a diminution in size of a small part of the lumbar grey matter on the left side, with a corresponding decrease in the number of the ganglion cells. The rest of the nervous system was quite normal. The latest pathological work in reference to this disease was by Von Babes, already mentioned in connection with the type just described. One of his cases was an example of Erbs paralysis, and he found in it the same nervous lesions as he found in the Pseudo-hypertrophic paralysis, under which heading they have been described; but, as already stated, his results have not as yet been confirmed.

The "Facio-scapulo-humeral"type of muscular atrophy was first recognised by Duchenne, but his observations were some time afterwards much amplified by Landouzy and Dejerine.(7) That they considered the type to be a pure myopathy is proved by the title they chose for their paper, "Myopathic atrophique progressive, Sans Neuropathie." In support of this view they were able to bring forward the evidence they obtained from an autopsy made in one of their cases, in which they found the nervous system perfectly normal. A year later they again had the opportunity of making a post mortem

examination on a case[8] they considered belonged clinically to this type, and in this also they found the nervous system, central and peripheral, unaffected. It is interesting to note, moreover, that although during life they could observe no changes in the muscles of expression, such as usually characterise this disease after death, they easily demonstrated that these muscles were atrophied, or beginning to atrophy. No further addition to our knowledge of this type of muscular dystrophy has been made of late years, so that, as far as we can say, it is, as its discoverers described it, a pure myopathy.

The "Hereditary form" of Leyden is very similar, in its clinical and pathological features, to Pseudo-hypertrophic paralysis. The condition of the muscles is the same, pseudo-hypertrophy occurring among the wasted muscles. The chief feature of the disease noted by Leyden is the strong hereditary disposition it shows. No fresh light has been thrown on this condition since it was first described.

The "Atrophic form of Myopathic Muscular Atrophy". Here we have a condition hardly to be distinguished from the above, except that examination of the few cases that have been noted reveals the fact that no

pseudo-hypertrophy has as yet been described. The pathology of the disease, as far as we know it, is the same as pseudo-hypertrophic paralysis and the Hereditary form; and of late years no researches into the cause of this disease have been published.

It is to be noted that many writers do not mention these two last forms as distinct types of muscular dystrophy, but their clinical and pathological features clearly assign them to that disease, and I think our present knowledge is not sufficient to consider them merely variations of Pseudo-hypertrophic paralysis.

The "Diffuse form of Muscular Atrophy." We are indebted for most of our knowledge of this rare form of muscular dystrophy to Doctors Thomson and Bramwell.(9) They have each published cases and Dr. Thomson has also given us the result of a post mortem examination made by Doctor Bruce. Clinically the disease resembles the other types of muscular dystrophy in a marked manner; a point worthy of note being that it is an affection of very early childhood. The results of Dr. Bruce's examination at the autopsy were briefly as follows:- The muscles resembled, in their microscopical appearances, muscles from cases of Erb's paralysis, the fibres being atrophied, nuclei being

increased, and vacuolation being present. But the muscles were not alone affected, the peripheral nerves also showed striking changes, by the presence of round and spindle cells between the nerve fibres, some nerve bundles were compressed and atrophied, and in some cases even, were replaced by fibrous tissue. In the spinal cord it was also noted that a number of cells in the anterior horn had disappeared, while those that were left were more or less atrophied. I can find no account of any other autopsy on a similar case to confirm these results, therefore they must represent the extent of our present knowledge of the matter.

The last of the types of Progressive Muscular Dystrophy, the "Peroneal type", was first recognised by Charcot and Marie[10] in France, and, about the same time and independently, by Tooth[11] in England. Tooth collected together the already published cases, and attempted to show that the disease was neuropathic in origin. His contention was supported by post mortem examinations made by Virchow,[12] Oppenheimer[13] and Friedreich.[14] These observers found interstitial neuritis in the nerves supplying the affected muscles. More recent writers are agreed that, clinically, the cases seem to be certainly due to a peripheral nerve

lesion; and autopsies by Dubreuilb, and Déjérine and Scoltas, confirm the results obtained by Virchow and the other earlier observers, interstitial neuritis being found; while in the case described by Déjérine and Scoltas there were changes in the posterior columns of the cord, probably secondary in character. Dr. Sachs,(15) in America, and Dr. Herringham,(16) in England, on the other hand consider rather that the disease has a central origin, that it is a myelopathy, but they can bring forward no pathological facts to prove their theory.

And now let us consider for a moment what all this means. It may be urged that it is unwise and useless to attempt to classify diseases on what may be, I think fairly, considered insufficient grounds. On the other hand if classification is attempted on broad lines alone, and considered merely as a provisional division of the subject, I think it would tend to advance rather than impede the study which will, in the end, demonstrate the true cause of the disease. And here I would enter a plea for the retention of the names given to the various types by their first describers, and adopted in the list given above. To my mind it is much to be regretted that some of the types

already possess dual titles. Nothing is gained by this, and it certainly promotes confusion. For instance in the "Twentieth Century Practice of Medicine" (17) Tooth's "Peroneal type" is found under the heading "Diseases of the Peroneal Nerve." It is certainly true that the disease, as a rule, commences by interstitial neuritis of that nerve, and atrophy of the muscles supplied by it, but Charcot and Marie have conclusively proved that the muscles of the hand and arm are very shortly afterwards involved, therefore the Ulnar and Median nerves &c. have a claim also to be included in the title, which would make it most cumbersome and ridiculous. Both Charcot and Tooth are agreed that the disease belongs of right to the muscular dystrophies, therefore why not leave it among them till such time as it is certainly shown to belong elsewhere. Any classification put forward just now must, of course, be a tentative one, and I think should be based entirely on our present pathological knowledge. That knowledge is, as far as it goes, positive, and by means of it we can trace connecting links between the various types, so that if, in the future, it is made clear that the above mentioned varieties of muscular dystrophy are in truth but clinical

variations of the same disease, no very marked changes in our consideration of the affection will be needed.

It is by no means certain that in the several types of Muscular dystrophy enumerated above, we may not have what are really unnecessary divisions. For instance Erb's "Juvenile form" and the "Facio-scapulo-humeral type" of Landouzy and Déjérine are in many points identical. The chief differences between them being that in the former disease the shoulder girdle muscles are first involved, and also there is occasional pseudo-hypertrophy of some of the muscles; while in the latter the face muscles are first implicated, and there is no pseudo-hypertrophy. This pseudo-hypertrophy of the „Juvenile form" connects it in a marked manner with ordinary Pseudo-hypertrophic Paralysis, which is, in turn, linked to the "Facio-scapulo-humeral type" by the latter's resemblance to the "Juvenile form" of Erb. Again the morbid processes in all three are, as far as have yet been ascertained, alike; they are all myopathies, and their clinical history and progress are very similar. It is worth remembering, also, thet Remak[18] has published a case which he considered to be one of Erb's paralysis, in which the face muscles were involved after those of

the shoulder girdle. In the case I am about to describe a similar state of affairs existed. This, in my opinion, tends to show how closely related are the types of Erb and Landouzy; nay! may they not be identical? Von Babes, too, at the post mortem examinations he made on a case of Pseudo-hypertrophic paralysis, and on one of Erb's paralysis, found, in both, similar nervous lesions. Although his results have not as yet been confirmed, they are certainly an additional proof of the close connection between the two types. Further, the types placed fourth and fifth on the list, the "Hereditary form" of Leyden, and the "Atrophic form of Myopathic Muscular Atrophy," are, in many points, alike, while the pseudo-hypertrophy in the "Hereditary form" unites them to Pseudo-hypertrophic paralysis, which they also closely resemble in their clinical aspects, as well as in the pathological changes found in the affected muscles; both conditions being, as far as it is at present known, like the above types, pure myopathies. But type number six has some very different characteristics. Cases of "diffuse muscular atrophy in children" seem to be very rare, but the condition has been described by Doctor Thomson and Doctor Bramwell. In Dr. Thomson's case the wasting

began very insidiously when the child was about 12 or more months old, and spread gradually, the lower limbs being the first affected, and then the upper limbs and neck. The wasting was quite symmetrical and there was never any hypertrophy or pseudo-hypertrophy. Post mortem Dr. Bruce found marked nervous lesions, and he considered the cause of the disease to be myelopathic rather than myopathic. It is clear, therefore, that this condition cannot be included in a classification under the same heading as the five previously mentioned types of myopathic atrophies, for it appears to be essentially a primary myelopathy.

Lastly we have left variety number seven, the "Peroneal type" of muscular atrophy. This also has certain strong characteristics which mark it off from the other types noted. Both clinically and pathologically, as far as can at present be judged, the disease seems to be caused by lesions in the peripheral nerves. The few autopsies that have been made clearly show this. There is no hypertrophy or pseudo-hypertrohpy to be found in the affected muscles, and no primary spinal changes have as yet been demonstrated. Yet there can be no doubt that the disease is a type of Progressive Muscular Dystrophy, a type separated

from the others by being neuropathic in origin.

In studying the characteristics of these various types in this manner there are some points, suggested by the symptoms and pathology of the "Diffuse form," which, in my opinion, are worthy of further consideration. Clinically this disease resembles the other types in a more or less definite way; Pathologically the condition of the muscles is similar to the condition found in the affected muscles of the other forms of muscular dystrophy, while the changes in the peripheral nerves are identical with those found in the "Peroneal form." So far this form may be said to act as a connecting link between what we at present call the pure myopathies and Tooth's neuropathic form. But we have still further nervous lesions in this "Diffuse type." The cells in the anterior horns of spinal grey matter are affected, and in this it differs from the other types in which no such change has been so far demonstrated. May not this difference be explained by the fact that this disease starts in infancy, when these spinal cells are in a state of growth and development, and so are more easily affected. Whereas the other types of muscular dystrophy are diseases of late childhood or youth, when the spinal

cells are in a more stable condition.

We have gone as far as our present knowledge can lead us, and further speculation is useless. We can, however, using a pathological basis, group the various types as follows:-

I. Myopathic Atrophies.

a. Pseudo-hypertrophic paralysis. b. Erbs "Juvenile form." c. "Facio-scapulo-humeral" type. d. Leyden's "Hereditary form." e. The "Atrophic form."

II. Neuropathic Atrophies.

a. "Peroneal type."

III. Myelopathic Atrophies.

a. "Diffuse form of Muscular Atrophy."

And now I wish to relate the history of two cases which afford a very striking picture of two different types of muscular dystrophy. On this account I think it wise to give the notes of the cases in full. The first case is that of a young woman who first noticed weakness and atrophy commencing in the shoulder girdle muscles. When she came under my care the muscles of expression were also involved. There was no affection of the lower extremities, no fibrillar tremor, and no

reaction of degeneration was made out. No history of similar cases in her family could be obtained. The second case is that of a young man who, while leading an active life, began to suffer from weakness in his legs. Atrophy of the Peronei and Calf muscles soon showed itself, and, later, the muscles of the hand were affected. Fibrillar tremors were observed, and the reaction of degeneration was obtained in some of the atrophied muscles. A maternal uncle suffered in a similar manner, and there is a marked history of neurosis in the family on the mother's side. I will consider the question of the diagnosis in these two cases in its proper place.

Sarah Bell, a young woman aged 22 years, first came under my observation on October 28th 1896. She was a worker in a cotton factory, fulfilling the duties of a "speed tenter." She has lived in Oldham (Lancashire) all her life. Her home surroundings have been most comfortable. The patient started work in the factory at the age of 13 years, and has continued in that employment until her present complaint put a stop to it.

Previous History.

The patient has had a remarkably healthy life. As a child she was very free from sickness, the only illness in childhood, that can be recalled, being measles. Her parents state that as regards teething, learning to walk, etc., they can remember nothing unusual; she was not a particularly fat child, but was always muscular and well developed. Her school-days passed uneventfully, and no information, shedding light on her present condition, can be obtained. At the age of 13 years she left school, and went to work in the cotton mill, at that time she always felt equal to her work, and was in no way fatigued by it. At the age of 15 years she suffered from pain in both knee joints. She can give no cause for this, her Doctor called it rheumatism, and a fortnight's rest sent her back to work quite well again. She has had no serious return of this pain, but the knee joints are still a little stiff. The patient says that at the age of 18 years she had to give up her work for a time, owing, as she puts it, to "general weakness." However she got stronger, and returned to work for a little over nine months, when she first noticed her present symptoms.

Her present illness began in this way. Towards the end of the year 1895 the patient was working as a speed tenter, she was then 21 years old. One of her duties was to place empty "bobbins" (for winding silk) on the spindles which turned them round. These spindles were placed about a foot above the patient's head, and she discovered that she was becoming unable to put the "bobbins" up slowly. The bobbins are quite light, and the patient says she used to jerk her arm up, and that if she did not get the bobbin into its place at once, the arm fell slowly down to her side again. Another thing she noticed was that she could no longer stand to "do up" her back hair, but had to sit and rest her elbows on the dressing table. She continued at her work thinking that the exercise would be good for her, but she made no improvement. She was under a Doctor's care for a time, but without benefit. In September of 1896 she took further advice, and was recommended to come into the Oldham Infirmary. She was admitted on Oct. 28th 1896, and at that time she had been aware of muscular weakness in the shoulders for 13 months.

Family History.

Careful and persistent inquiries fail to bring out any evidence of either muscular weakness or paralysis in the family. Most of its members have enjoyed long lives, and no hereditary disease is present. The patient's father and mother are both living. The father a strong, healthy, well made working man; the mother only troubled with "rheumatic" pains in the left knee. The patient is the second child in a family of eleven, 8 brothers all living and well, and two sisters also in good health.

Present Condition.

As the patient stands before you it is noticed that she is of medium height, (five feet one inch), and generally seems well nourished. Her expression is somewhat vacant, and her lips thick. Her gait presents no unusual features, and she is quick and active on her feet. Her attitude is not abnormal in any way. She has no difficulty in rising from the recumbent to the erect posture. But when the clothing is removed from the arms and trunk, some very striking features are revealed. It is at once seen that her shoulders are very sloping, the neck very broad, and the scapulae very prominent. In contrast to the centrally prominent

No. 1.

Case of "Sarah Bell" Erbs Paralysis.

Showing general appearance - the thick lips, broad neck, sloping shoulders and wasted condition of the Pectoral Muscles and the Deltoids.

deltoid muscles, and well developed forearm and hand muscles, are the weak looking upper arms. The lower extremities are strong and well formed. These characteristics are shown in the first of the series of photographs I have taken of the patient.

Detailed examination of the muscles.

I. Of the Trunk and Upper Extremities.

The Pectorals (major and minor) are gone on each side except a thin layer of muscular tissue, the remains of the inferior border of the Pectoralis major; this remains on both sides.

The Trapezii can barely be made out.

The Latissimi have disappeared on both sides, and the same can be said for the Serrati magni.

The Rhomboids are greatly atrophied on both sides.

The Levator Anguli Scapulae seem normal on both sides, as also the Supra spinati.

The Infraspinati on both sides are markedly atrophied.

The Teres major and minor on both sides, as also the Subscapularis, are apparently normal.

The Recti, and abdominal muscles generally, present no unusual features.

The Erector spinae on both sides are healthy.

The Deltoids present certain well marked features, uniform on the two sides. The Anterior and Posterior portions of the muscle are extremely wasted, while the Middle portion, on the contrary, is powerful and somewhat hypertrophied, though this may only be a seeming hypertrophy, from contrast with the surrounding atrophied muscles.

The Biceps on both sides are flabby and weak, while the Triceps of both arms are greatly atrophied.

The muscles of the Forearms and Hands are well developed and powerful.

The wasted muscles feel soft and flabby, while the middle portion of the Deltoid, and the muscles of the Forearms and Hands, are firm. There are no fibrillar twitchings, the mechanical irritability of the affected muscles is lowered, and, in the case of the Pectorals and Latissimi, absent altogether. Their behaviour to the electric current, either Galvanic or Induced, is the same, that is to say, in the atrophied muscles there is only a very slight reaction to even strong currents. The reaction of degeneration was never observed, although the muscles were tested carefully many times. The skin reflexes in the upper extremity and trunk are normal, but the Tendon reflexes are absent.

II. Of the Head and Neck.

The Frontals, the Orbicularis palpebrarum, and the Zygomatics, are all normal, as also are the Ocular muscles and the Pupils. The muscles of the tongue, of mastication, and of the pharynx, are all perfectly healthy. But the Buccinators are distinctly weak. The patient's lips, as before noted, are very thick, and she is quite unable to whistle or blow out her cheeks. Again when she attempts to smile the angles of the mouth are not drawn upwards, but laterally outwards (rire en travers). There is also distinct wasting on each side of the chin. The patient cannot say when this wasting commenced, as, until her attention was drawn to it by the examination I made, she was not aware that there was anything peculiar in her expression or in the movements of her lips.

Of the muscles of the Neck the Sterno mastoids alone present features worthy of note. On both sides these are seen standing out prominently, and they are firm and hypertrophied.

III. Of the Lower Extremities.

In the lower extremities there is no marked enlargement of the muscles, and there is no wasting. All the movements performed by the muscles of the legs

No. II.

Case of "Sarah Bell" Erbs Paralysis.

Showing "dislocation" of scapula,
due to atrophy of Serratus Magnus
and the Rhomboids.

are done powerfully. There is no increased mechanical irritability, and their reaction to the Electric Current is normal. The skin and tendon reflexes are present, but in no way exaggerated.

If the arms are stretched out in front of the patient, both scapulae are "dislocated" backwards, and stand out like wings; and then, on moving the arms slightly up and down, the scapulae and humeri seem to move as one. This condition is well shown in the accompanying photograph. Also, as shown in the next photograph, if the hands are placed beneath the armpits, and the patient lifted, she has no power of fixing the shoulders, so they go up towards the ears, and the patient feels to be slipping through your hands. The first of these conditions, namely the wing-like projection of the scapulae or "scapulae alata," is due to absence of the Serrati magni and the Rhomboids; while the loss of the power of fixing the shoulders against resistence is due to atrophy of the Pectorals and the Latissimi.

Again, if the arms are abduced at the shoulder to the horizontal level and then brought down strongly to the side against resistance, the scapula is drawn towards the upper arm, and its angle is turned outwards,

No. III.

Case of "Sarah Bell" Erbs Paralysis.

Patient cannot fix the shoulders against resistence - due to atrophy of the Pectorals and the Latissimi.

showing that the Trapezii and Rhomboids are unable to fix it.

Otherwise the patient is perfectly healthy. She is an intelligent girl, with a good memory and mental development.

There are no vaso motor or trophic disturbances, save a slight tendency to blueness and coldness of the hands. The functions of the bladder and rectum are unaffected. There is no impairment of the sensibility of the skin to tactile impressions, heat, cold, and electrical stimuli. The special senses are as usual. The condition of the Alimentary, Respiratory, Circulatory, and Genito-Urinary systems calls for no remarks. The patient may therefore be said to be in"good condition" save for the muscular atrophy and its attendant deformities.

Now to consider the diagnosis of the disease. And in the first place I think it is fair to conclude that the condition is a primary muscular atrophy. That it is not a Progressive spinal muscular atrophy is certain, because, in that disease, the wasting almost always begins in the small muscles of the hand, which in this case are unaffected; and, again, in spinal muscular atrophies, fibrillar twitchings are

present, electrical irritability is long preserved, the "reaction of degeneration" in the affected muscles can usually be obtained, and the tendon reflexes are not lost till late in the course of the disease; whereas, in this patient, the reverse of these facts are found. That the wasting is not due to Syringomyelia is shown by the absence of pain, anaesthesia, and alteration in the temperature of the affected parts; also, in that condition, the "reaction of degeneration" occurs in the wasted muscles. Peripheral Neuritis (multiple) is excluded by the absence of any cause in the history of the case for such a condition, as lead or alcohol poisoning, or any severe illness such as Typhoid, Diphtheria, etc., preceding the onset of the symptoms. And, again, the absence of pain, sensory disturbances, and "drop wrist," put Peripheral Neuritis out of court. The atrophy of the muscles cannot be secondary to any disease of the shoulder joints, as careful examination and the history fail to produce any evidence of such a condition. Lastly, we have to exclude the later stages of Pseudo-hypertrophic paralysis, and I think this may be done from the fact that the patient has never had any weakness or enlargement of the muscles of the legs, that she

first noticed the shoulder muscles to be affected, and that her childhood and early youth were free from such troubles.

Therefore we may say that this is a case of muscular atrophy occurring in a girl about twenty years of age, without any discoverable cause. That the atrophy was confined, in a peculiar manner, to the muscles of the neck, shoulder girdle, and lower part of the face. That this atrophy progressed. That, as a result of it, certain characteristic signs of "juvenile muscular atrophy" were produced, especially loss of power to fix the shoulders and scapulae against resistence; signs that Erb considered of the greatest diagnostic importance. That the examination of the sensory functions revealed no pathological changes in them; and lastly, although there is no family history obtainable in the case, and heredity seems to play usually an important part in the causation of the disease, we cannot say that similar cases have not occurred in the family before, but only that they have not been observed. On these grounds then I think it may be concluded that the case is one of Juvenile Muscular Atrophy. It may be added that, in this case the muscles of the lower part of the face are involved,

but as the girl first noticed the shoulder girdle muscles to be weak, and knew nothing of the atrophy of the face muscles till questioned about them, I do not think there is sufficient evidence to stamp this as an example of the Facio-scapulo-humeral type, but prefer rather to consider it a case of "Erbs juvenile muscular atrophy," with affection of the face muscles.

Treatment and Progress of the case.

A tonic, containing arsenic and Strychnine, was prescribed, and a generous diet ordered. Locally, gentle massage of the affected muscles was performed twice a day, the sittings being short; weak Faradic currents were also applied. At the end of a fortnight of this treatment the patient expressed herself as feeling stronger. The movements at the shoulder joints were decidedly more vigorous, and the girl was able to raise her hands to the top of her head. The treatment was persevered with for ten weeks, when the patient decided to go home. At the end of this time there was marked improvement in certain of the wasted muscles. The anterior portion of the Deltoids, the Trapezii, and the Rhomboids, had decidedly increased in size and power, and the patient was able to do up

her hair. No improvement could be found in the Pectorals, Senati, or Latissimi. Her general health was good, and she arranged to come up frequently, as an outpatient, for the massage and battery. She left the hospital on January 11th 1897, and attended the outpatient department, regularly, for about six months, during which time her condition remained the same as at the time of discharge, there being no further improvement. Then, for a time, I lost sight of her; however she came to see me again during the first week of January 1898. I had not seen her for over six months, and during that time she had received no treatment. Her condition (January 1898) was worse than when I first saw her in October 1896. She has lost all power in the shoulder muscles, so that the arms cannot be raised to the head, or even to the horizontal level. There is some slight lordosis in the lumbar region. The muscles of the forearm and hand are still unaffected, and she is able to work a knitting machine. The lower limbs also remain quite healthy, and she would enjoy walking exercise but that the slightest hurry or exertion causes her to become very breathless, due no doubt to the atrophy of the muscles of forced respiration. For the rest the patient is in the same condition as when first seen, and continues to enjoy moderate health.

Case II.

James Button, aged 23 years, first came under my care on October 13th 1897. His employment was that of a mechanic in a large iron works. He has been in constant work since the age of 10 years, and, until recently, he has had no difficulty in fulfilling his duties. He is a man of regular and steady habits; a moderate smoker, smoking a light tobacco in a pipe; and, although not a total abstainer, only drinks a small amount of alcohol, and that in the form of beer. The patient was born, and has spent the most part of his life, in Oldham (Lancashire), however, during the past year, he was a considerable time in North America.

Previous History.

In infancy, the patient's mother tells me, he was treated by a Doctor for rickets, and he was at that time slightly bow-legged. He recovered completely. He is said to have commenced to walk when 18 months old, and his mother says that in childhood he was just as other children. At the age of four the patient had measles, and at the age of six scarlet fever, but recovered from both in every respect. When he was about ten years old he commenced to learn his trade,

that of a mechanic. He is chiefly employed in putting up machinery for cotton spinning, and he says that the work is not heavy, and that until recently he has always been able for it. In July 1896 he went over to America with some machinery that was to be erected there, and remained there some time, doing his work in various parts of the Continent. Early in April 1897 he noticed that walking was a trouble to him, and that he tired sooner than usual; also that, occasionally, his left foot turned in so that he trod on the outside of it. The weakness in his legs increased so that his gait became noticeable, the most difficulty being experienced in going up steps. In the early part of June 1897 he noticed that he had partially lost the use of his toes on the left foot, walking had become more difficult, and his gait less steady, necessitating the constant use of a stick. He saw a Doctor, who said he had "spinal disease," but gave no further particulars. He remained in America until July 11th 1897, when he sailed for England; the weakness in the muscles of his legs had steadily increased, but, by the aid of two sticks, he could still get about, and managed to walk a little on the ship. He arrived in Oldham on July 20th 1897 feeling much better generally for the voyage,

he also says he thinks he could walk a little better. He remained at home, walking very little and with great difficulty; he now noticed that he had "cramps" in the legs, often coming on when in bed at night, keeping him awake. On two occasions, when walking in the street, he fell, the first time because his"foot turned under him," and the second because his "knees gave way." Towards the end of September 1897 he noticed that there was some wasting in the muscles of his left hand, and that he could not use it as well as formerly. He put himself under a Doctor's care, and after a time was recommended to come to the Infirmary in Oldham for treatment. The patient does not seem to have had syphilis; although he admits the possibility of infection; there is no history or other evidence of its having occurred.

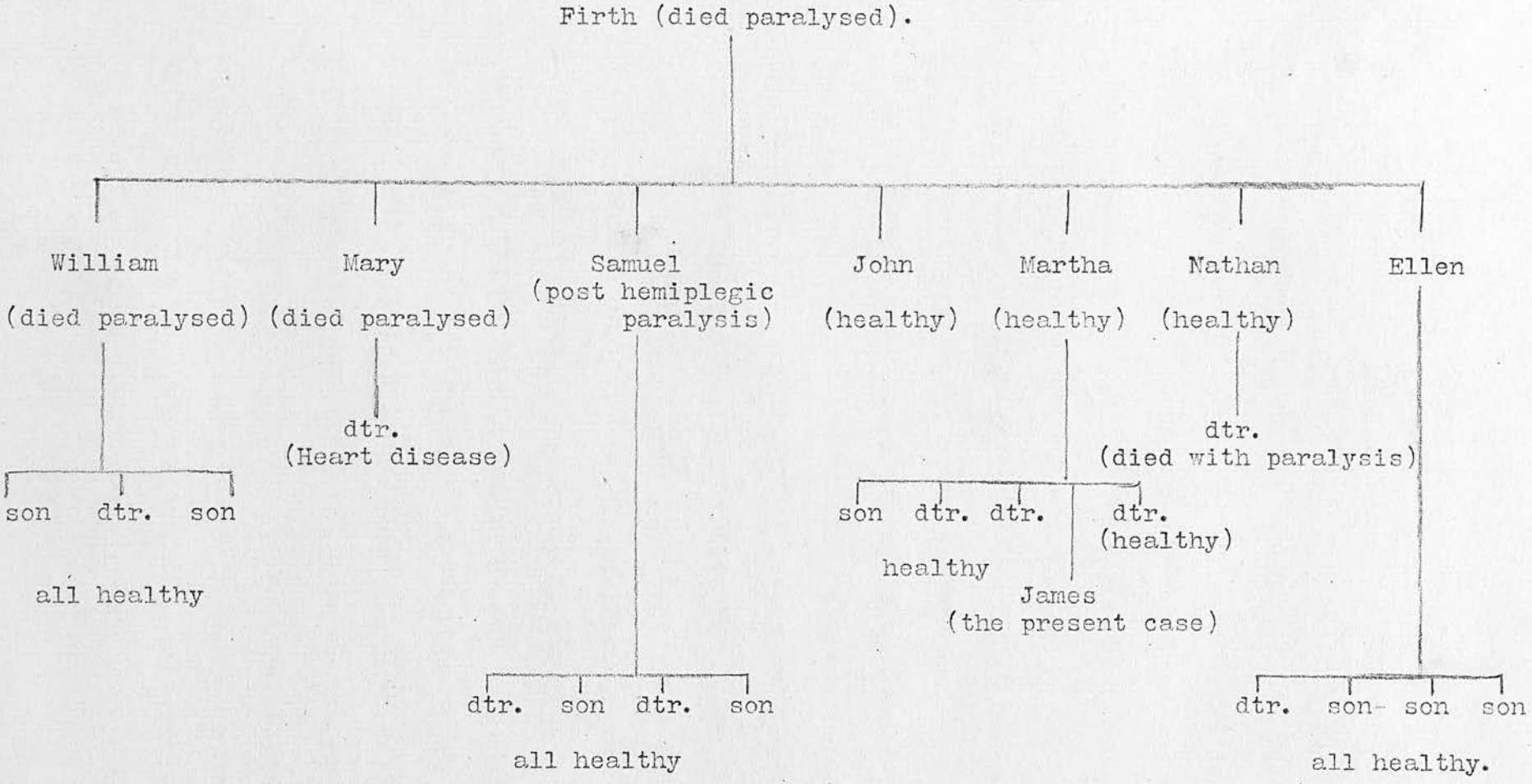
Family History.

The family history is of extreme interest, as, although no actually similar cases have as yet occurred, there is marked history of neuroses on the mother's side of the house. The patient is one of a family of five, one brother and two sisters older than himself, and one sister younger. They are all perfectly

well, and show no signs of muscular wasting. His father has long been a delicate man, suffering chiefly from derangements of the stomach, but has had no paralysis. His mother is a very healthy woman.

His maternal grandfather suffered from muscular weakness and paralysis. The disease commenced when he was about sixty years old, and first attacked the legs. He used to fall without any seeming cause, and on two occasions broke his leg. No very marked atrophy of the leg muscles was noticed, but they were extremely weak. The disease spread into his arms, and, at the time of his death, the arms were quite powerless. His mind was clear, and his speech remained unaffected.

This man had a family of seven children, four sons and three daughters, the present patient's uncles and aunts. The following is a short account of their several conditions.


Firth (died paralysed).
William
(died paralysed)
son
dtr.
son
all healthy
Mary
(died paralysed)
dtr.
(Heart disease)
Samuel
(post hemiplegic
paralysis)
dtr.
son
dtr.
son
all healthy
John
(healthy)
Martha
(healthy)
son
dtr.
dtr.
dtr.
(healthy)
healthy
James
(the present case)
Nathan
(healthy)
dtr.
(died with paralysis)
Ellen
dtr.
son
son
son
all healthy.

The eldest son, William, was a clerk in the Lancashire & Yorkshire Railway Co's employ. In March 1875 when 42 years of age he had a fall from a train. He was not severely hurt, and was able to walk home, but in the course of a few weeks his legs became weak, the muscles began to waste, and, a little later, the muscles of his arms began to do the same. The muscles wasted rapidly, and he became a mere skeleton, so much so that one of his brothers could carry him about he being a man of six feet in height. He died at the end of the same year, 1875, and it is said that, towards the end of his illness, his speech became very indistinct. He left a family of two sons and one daughter who are all healthy.

The second child, Mary, died, aged 42 years, of paralysis. The wasting began in the thumb of her left hand two years before her death, and spread up her left arm. The muscles of the left leg then became affected, and the disease is said to have been confined to the left side of the body. There is no history of an apoplectic stroke. She had one child, a daughter, now living who has heart disease but otherwise is healthy.

The third child, Samuel, is still living and is 60 years old. He has some paralysis of the left arm and leg, which appears, from the history, to be the result of Apoplexy. He has a family of two sons and two daughters all healthy.

The fourth child, John, is living and well.

The fifth child, Martha, is the mother of the patient, she herself is a healthy woman, and the rest of her children are free from disease.

The sixth child, Nathan, is also living and healthy. He had one child, a daughter, who died about 14 years ago at the age of 12 years. This child died paralysed, but the history of the illness is very vague. The girl is said to have had a "stroke" which left her paralysed, and some months later a second, and then a third, from which she died.

The youngest child, Ellen, is still living and has a healthy family, consisting of three sons and one daughter.

Present Condition.

The patient is a thin pale faced man, 5 feet 8 ins. in height and 9 stone 8 lbs in weight. There is nothing in his expression to attract attention. When standing in the erect posture the patient is

perfectly able to maintain his balance. There is some curving forward of the spinal column in the lower dorsal and lumbar regions. The feet are placed somewhat apart and the toes point straight forward. Very little force is required to disturb the patient's equilibrium when standing erect, and if he is given a slight push he often is unable to recover his equilibrium and falls to the ground. His gait is peculiar, the knees are raised higher than usual to keep the toes off the ground, and when the step is made the balls of the toes touch the ground first. The feet are placed flat upon the ground. Also it is to be noted that there is no irregular throwing about of the feet but the foot points straight forward as in health. His method of rising from the supine position on the floor is to turn over and get on his hands and knees, he then gets his feet onto the ground and climbs up his thighs with his hands, but is often unable to get into the erect posture without assistance. He has the power of fixing the scapulae to the thorax.

Motor Power.

All the movements of the upper and lower extremities can be performed, save that the patient cannot move the toes of either foot. In the upper extremities

the movements at the shoulder, elbow, and wrist, are performed powerfully, but the movements of the fingers of the left hand are very weak and uncertain, and his grasp is feeble, while in the right hand the movements of the fingers are powerful. In the lower extremities he has fair movements of the hips and knees, at the ankles the movements are weaker, and as stated above he is unable to move his toes.

The Condition of the Muscles.

I. Of the Face and Neck.

There is no wasting, or loss of power, in any of the muscles of the face or neck. The muscles respond freely to electricity.

2. Of the Upper Extremities.

In the right arm there is no muscular atrophy or hypertrophy to be found. When the arm is at rest no "drop wrist" or other deformity is to be observed. The muscles are firm to the touch, and contract powerfully. There are some slight tremors of the fingers, but no fibrillary twitchings were noted. The muscles react well to both Faradic and Galvanic currents, except the muscles of the Thenar and Hypothenar eminences; in these, although no diminution in size

could be made out, there was a very poor reaction to either current.

The left arm for the most part resembles the right. No atrophy or hypertrophy present in the upper or forearm, but the Thenar and Hypothenar muscles are markedly wasted, as are also the Interossei. When the limb is at rest there is a decided tendency for the wrist to be held in an extended position. There are no fibrillary twitchings. All the muscles of this arm react well to electricity except the small muscles of the hand, and in these even very strong currents produce no contraction. There is no reaction of degeneration.

3. Of the Trunk.

The muscles of the thorax and abdomen present nothing to note.

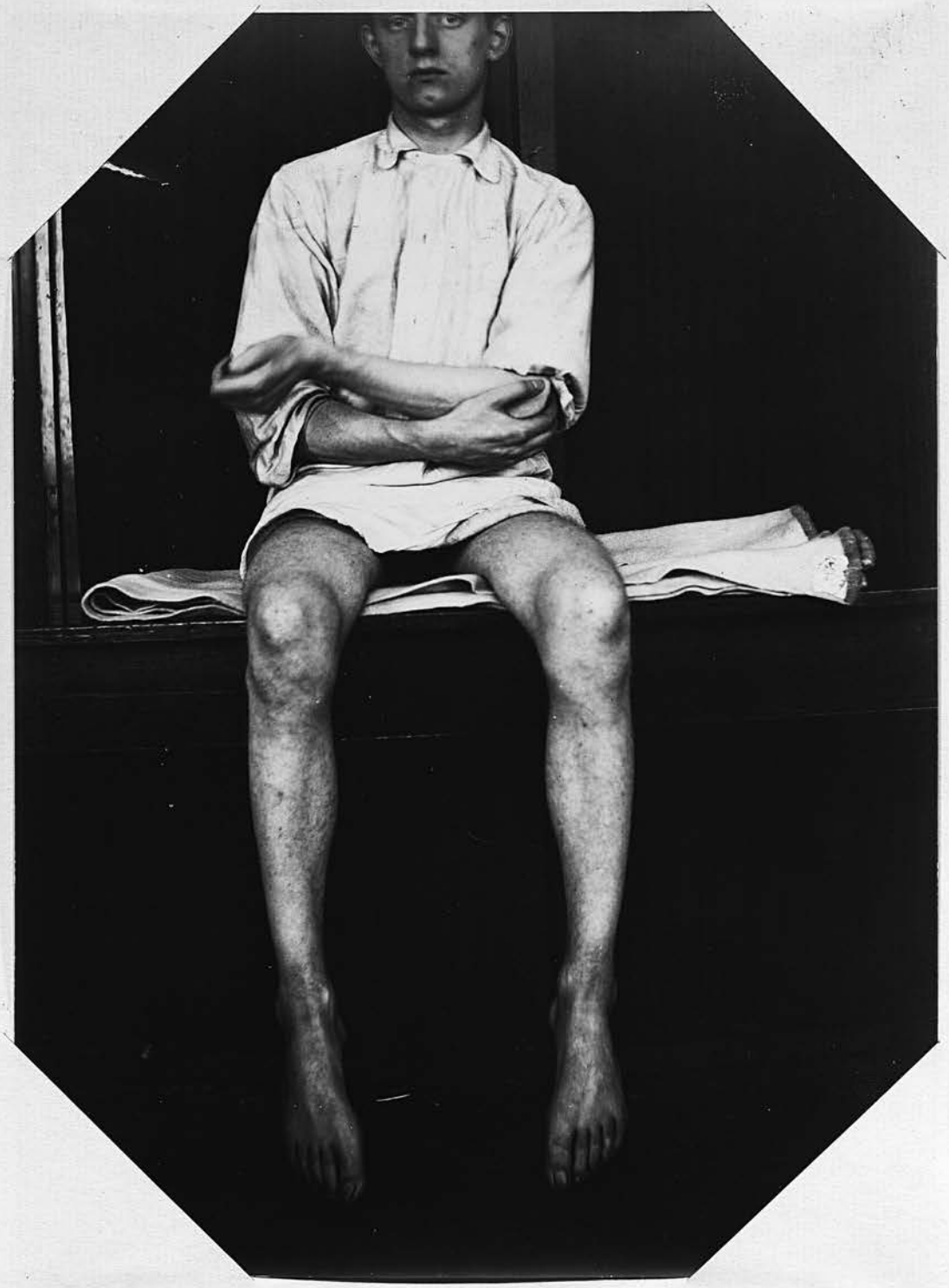
4. Of the Lower Extremities.

As the patient lies in bed the appearance of both legs is the same. The muscular development of both limbs is very poor. The feet are somewhat arched, and the insteps are high, but there does not appear to be any proportionate shortening of the foot. The first phalanx of all the toes is bent upwards, and

the second downwards. The heels are slightly drawn up, the soles of the feet are turned inwards, and their inner borders are slightly lifted, thus increasing the concavity on the inner side of the foot. On both sides the Extensor Brevis Digitorum is markedly atrophied, also the Tibialus Antichus, while the Peronei cannot be made out at all. The shaft of the Fibula is easily felt along its whole extent. The calf muscles ars amall and flabby, and do not contract strongly. The flexors and extensors of the thigh are small and weak, and there is no enlargement of the Gluteal muscles. Some fibrillary twitchings are seen in the atrophied Tibialus Antichus on both sides, also in the Extensors of the thigh. There is no reaction to either the Galvanic or Faradic currents in flexors of the ankle, the peronei, or the extensor brevis digitorum, that is, in the obviously wasted muscles; while there is only a very poor reaction in the flexors of the ankle and the extensors and flexors of the thigh. The reaction of degeneration was obtained in most of the wasted muscles.

I have taken several photographs of the patient, but unfortunately these do not bring out the marked wasting in the groups of muscles above mentioned as well as

No. I.



Case of James Button. "Peroneal type"

Showing general wasting of the leg muscles, the extreme extension of the ankle (patient cannot dossiflex his foot), and the position of the hand and wrist when at rest.

No. II

Case of James Button "Peroneal type"
Showing wasting of muscles in the leg, and the position of the foot when at rest, the heel drawn up, the sole turned inwards, and the inner border of the foot slightly raised.

No. III.

Case of James Button "Peroneal type"

Same as No. II only photograph taken from outer side of limb. Shows clearly the position of the toes when the limb is at rest.

No. III.

Case of James Button "Peroneal type"

Same as No. II only photograph taken from outer side of limb. Shows clearly the position of the toes when the limb is at rest.

No. IV.

Case of James Button "Peroneal type"

Shows wasting of the Peroneal muscles, also those of the leg generally. The outline of the Fibula can be clearly seen along its whole course.

could be wished. The notes beneath each photograph mention the most important points that are brought out by the picture.

The Reflexes.

The Plantar reflex is gone on both sides. The cremasteric and other superficial reflexes of the body are present. There is no ankle clonus, and the knee jerks are absent. The Triceps jerk is present in both arms. The organic reflexes are perfectly normal.

The Vasomotor and trophic condition of the skin.

The hands and feet tend to be blue and cold, and there is, at times, some venous mottling of the skin, especially of the feet.

Sensory functions.

The sensibility of the skin to tactile, painful, and thermal impressions is quite normal. The sensibility to electrical stimuli appears to be normal. The special senses are normal.

Mental functions.

Intellectually the patient is certainly up to the normal standard. He reads and writes well, has a

good memory, and takes a lively and intelligent interest in the progress of his malady.

There is nothing to note regarding the Circulatory, Respiratory, or Alimentary systems. The urine has been never found to contain albumen and is quite normal. Except the muscular weakness, occasional cramps in the legs, and some twitchings of the fingers, the patient has nothing to complain of.

In coming to a diagnosis on this case we have unfortunately, from a scientific point of view, to trust entirely to clinical evidences. These, however, I think are clear and well defined.

Here is the case of a young adult, leading an active life, who suddenly finds that his legs are becoming weak. Then marked wasting of the muscles is apparent, this wasting first showing itself in the peroneal and calf muscles. At the same time fibrillar tremors are observed in these muscles, and the "reaction of degeneration" is obtained in them. The disease spreads, the muscles of the thighs are affected, and later the small muscles of the hands. Lastly there is a strong "nervous" taint in the family, an uncle indeed, after, what at the time was considered a slight accident, suffered from what seems to have been

an exactly similar condition. These characteristics comply with those that Tooth formulated as the necessary symptoms of the "Peroneal type," and therefore I think the case to be an undoubted example of that form of "Muscular Atrophy." A symptom which struck me in this case, and which is mentioned in all the published cases of "Peroneal atrophy," is the gait. The patient walks with a peculiar "high stepping" action due to his being unable to dorsiflex the ankle, and so clear the ground with his toes. He has instead to raise the whole foot clear of the ground by means of his quadriceps extensor. I think, in view of the fact that this gait has been noted in all the cases, it may be regarded as a clinical symptom of great importance.

Progress of the Case.

The wasting in the muscles slowly advanced. The thighs, especially, becoming much weaker, so that the patient can no longer walk, or even stand, without help. The wasting also in the muscles of the hand is advancing, and the power of making fine movements with the fingers is lost. The patient's general health also is not so good, and he is gradually becoming more and more helpless.

A few words more in conclusion. Strümpell has suggested that all forms of muscular dystrophy "depend on a congenital tendency of the nervous system to degenerate." If this be true, the form that this degeneration takes has in many cases not yet been made out. It is of the highest importance, for the clearing up of this subject, that, in the future, no opportunity should be missed for making post mortem examinations on cases of this kind, special attention being given to the study of the peripheral nerves and spinal cord. Dr. Ferrier[19] has pointed out that certain levels of the cord contain groups of cells corresponding to groups of muscles. These cells form centres for the control of combined movements of these muscles, similar to those existing in the cortex of the brain. It may be that in some lesion of these cells lies the explanation of the clinical manifestations found in the muscular dystrophies. In all the various types groups of muscles are involved, and these groups are controlled by those spinal nerve cells. The theory that I would put forward is, that in the muscular dystrophies there is an hereditary tendency for these "control" cells of Ferrier to degenerate; but we must wait with patience for post mortem examinations to

demonstrate that this or some similar change is the true explanation of the disease. It is to be hoped that, in the near future, our knowledge of this most interesting group of nervous diseases will be much more definite; that we shall know if their pathology be the same or whether we have as great variations in their pathological as in their clinical manifestations, and here I must leave the subject. In conclusion let me apply the words of the Greek Poet to the views brought forward in this thesis.

"These opinions have seemed to me
to resemble the truth."

Nathaniel Caine.

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